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Focal Dermal Hypoplasia (Goltz Syndrome) Associated With Intestinal Malrotation and Mediastinal Dextroposition

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Focal dermal hypoplasia (Goltz syndrome) is a rare syndrome comprising developmental anomalies of tissues and organs of mesoectodermal derivation. We report on a characteristic case of focal dermal hypoplasia with the previously unreported association of mediastinal dextroposition and intestinal malrotation. © 1996 Wiley-Liss, Inc.

KEY WORDS: focal dermal hypoplasia syndrome, intestinal malrotation, mediastinal dextroposition

INTRODUCTION

Focal dermal hypoplasia (FDH) was first characterised in 1962 [Goltz et al., 1962]. The following year an extensive review identified 11 previously described cases [Gorlin et al., 1963]. The most recent and comprehensive review estimated that over 200 cases are known [Goltz, 1992]. The cutaneous manifestations are emphasised in most reports, hence the name; however, there are well-recognised associated anomalies of the eyes, skeletal system, and renal tract. We report on a girl with many characteristics of FDH but with the additional, previously unreported finding of mediastinal dextroposition and intestinal malrotation.

CLINICAL REPORT

The proposita initially presented at 20 weeks of gestation when routine antenatal ultrasound scanning demonstrated an omphalocoele. Cytogenetic analysis of amniocentesis fluid showed a normal 46,XX karyotype. The parents were healthy and non-consanguineous and there was no teratogen exposure. The primigravid mother was normal, 29 years old; the father was normal and 30 years old. This was also his first child. Later in the pregnancy bilateral hydronephrosis was noted; otherwise gestation advanced without complication and the proposita was delivered vaginally at 37 weeks of gestation with a birth weight of 3.25 kg.

The omphalocoele was repaired neonatally. At operation, intestinal malrotation was noted and subsequently confirmed by barium examination (Fig. 1). Facial anomalies (Fig. 2) included strabismus, displaced left lacrimal punctum, epicanthic folds, left incomplete cleft lip, a low cup-shaped left ear, bilateral pre-auricular sinuses, and a narrow nasal bridge with a broad nasal tip. The cutaneous manifestations were particularly

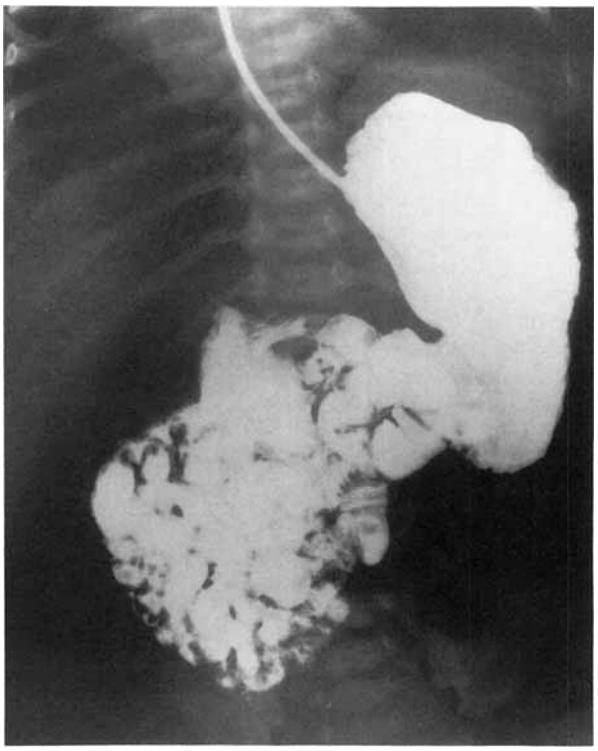

Fig. 1. Barium study demonstrating malrotation of the entire small intestine.

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Fig. 2. Frontal view of patient. Note strabismus, left incomplete cleft lip, left low cup-shaped ear, epicanthic folds, and broad nasal tip.

Fig. 4. Haematoxylin and eosin, ×60. Marked dermal hypoplasia with adipose tissue high in the papillary dermis.

striking, comprising linear striate lesions of fat herniation over the legs in the distribution of Blaschko's lines (Fig. 3) and focal atrophy-like cutaneous depressions over the arms and legs, again in a linear distribution. Biopsy taken from a representative area of the atrophy-like lesions demonstrated a markedly hypoplastic dermis with adipose tissue present high in the papillary dermis (Fig. 4). The linear fat herniations had almost completely regressed with minimal scarring by 3 months (Fig. 5). Bilateral clinodactyly of the fifth fingers was observed with bilateral syndactyly of the second and third toes. Toenails were absent from all ten toes; the fingernails were dystrophic.

Chest roentgenogram demonstrated mediastinal dextroposition with a small area of congenital emphysema in the right upper lobe (Fig. 6). Ultrasound study confirmed mediastinal dextropositon and detected a small, non-haemodynamically compromising patent ductus arteriosus. Bilateral non-obstructive hydronephrosis was demonstrated on abdominal ultrasonography. Further intestinal investigation demonstrated duodenal atresia, which was repaired subsequently and a Meckel's diverticulum. The patient has had par-

Fig. 3. Linear striate lesions of fat herniation over the left foot (age 10 days).

Fig. 5. Left foot (age 3 months), the same area as in Figure 3. The linear striate lesions have healed. Note syndactyly of the second and third toes.

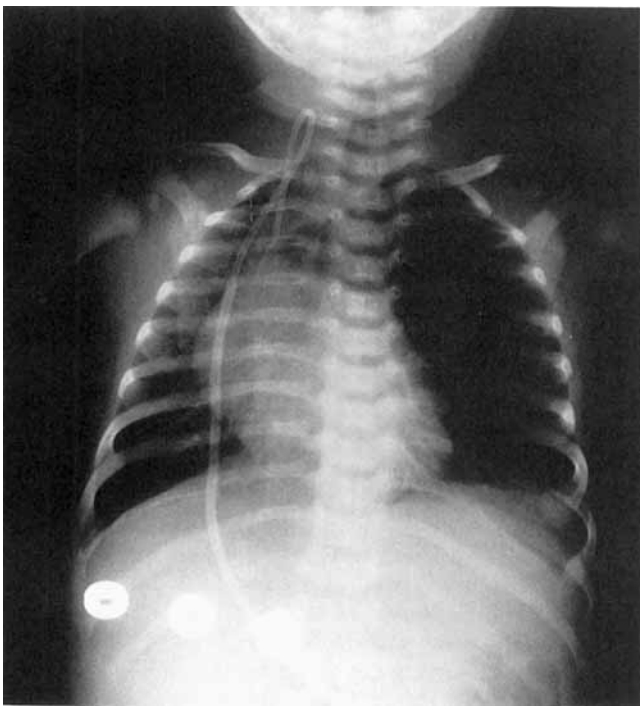

Fig. 6. Chest roentgenogram (age 10 days).

ticular difficulty with oesophageal reflux resulting in failure to thrive and requiring frequent hospital admissions for nasogastric and gastrostomy tube feeding. Recurrent urinary tract infections secondary to vesicoureteric reflux have necessitated bilateral ureteric re-implantation at 7 months.

X-chromosome inactivation studies were undertaken on cultured fibroblasts from the mother and proposita. The proposita demonstrates a markedly skewed, nonrandom pattern of inactivation; the mother demonstrates a normal, random pattern of X chromosome inactivation.

DISCUSSION

We report on a girl with minor facial anomalies, cutaneous ocular and skeletal lesions characteristic of FDH [Temple et al., 1990; Ishii et al., 1991]. X chromosome inactivation studies on the mother and proposita are strongly suggestive of an X-linked disorder. This is the first reported case of mediastinal dextroposition and intestinal malrotation in association with focal dermal hypoplasia.

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